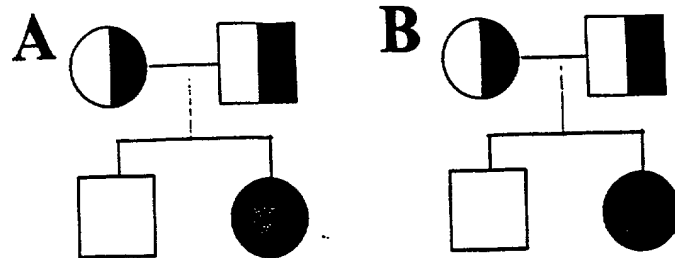


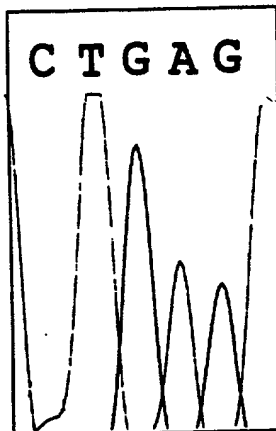
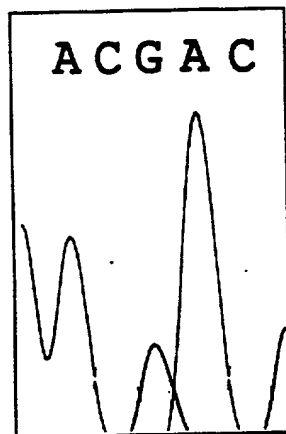
**Fig. 1**



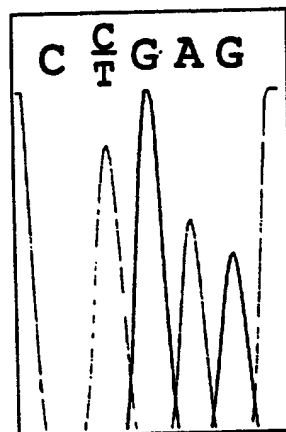
**Fig. 3**



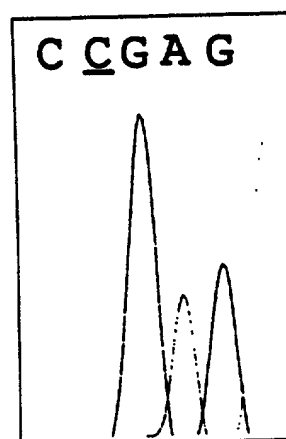
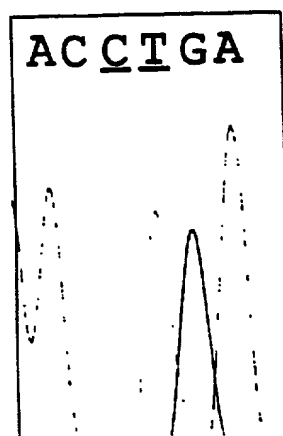
**Patient**



**Carrier parent**



**Healthy sibling**



1 MALGTTLRAS LLLGLLTEG LAQLAIPASV PRGFWALPEN LTVVEGASVE  
 51 LRCGVSTPGS AVQWAKDGLL LGPDPRIPGF PRYRLEGDPA RGEFHLHIEA  
 101 CDLSDDAEYE CQVGRSEMGP ELVSPRVILS ILVPPKLLLL TPEAGTMVTW  
 151 VAGQEYVVNC VSGDAKPAPD ITILLSGQTI SDISANVNEG SOQKLFTVEA  
 201 TARVTPRSSD NRQLLVCEAS SPALEAPIKA SFTVNVLFPP GPPVIEWPGL  
 251 DEGHVRAGQS LELPCVARGG NPLATLOWLK NGQPVSTANG TEHTQAVARS  
 301 VLVMTVRPED HGAQLSCEAH NSVSAGTQEH GITLQVTFPP SAIILGSAS  
 351 QTENKNVTLS CVSKSSRPRV LLRWLWGRQ LLPMEETVMD GLHGGHISMS  
 401 NLTLFARRED NGLTLTCEAF SEAFKETFK KSLILNVKYP AQKLWIEGPP  
 451 EGQKLRAQTR VRLVCLAIGG NPEPSLMWYK DSRTVTESRL PQESRRVHLG  
 501 SVEKSGSTFS RELVLVTGPS DNQAKFTCKA GQLSASTOLA VQFPPTNVTI  
 551 LANASALRFG DALNLTCSVV SSNPPVNLWS DKEGERLEGV AAPRRAPFK  
 601 GSAAARSVLL QVSSRDHGQR VTCRAHSAEL RETVSSFYRL NVLYRPEFLG  
 651 EQVLVVTAVE QGEALLPVSV SANPAPEAFN WIFRGYRLSP AGGPRHRILS  
 701 SGALHLWNVT RADDGLYQLH CQNSEGTAEA RLRLDVHYAP TIRALQDPTE  
 751 VNVGGSVDIV CTVDANPILP GMFNWERLGE DEEDQSLDDM EKISRGPTGR  
 801 LRIHHAKLAQ AGAYQCIVDN GVAPPARRLL RLVVRFAPQV EHPTPLTKVA  
 851 AAGDSTSSAT LHCARGVPN IVFTWTKNGV PLDLQDPRTY EHTYHQGGVH  
 901 SSLTIANVS AAQDYALFTC TATNALGSDQ TNIQLVSISR PDPPSGLKVV  
 951 SLTPHSVGL E WKPGFDGGLP QRF CIRYEAL GTPGFHYVDV VPPQATTFTL  
 1001 TGLQPSTRYR VLLASNALG DSGLAOKGTQ LPITTPGLHQ PSGEPEDQLP  
 1051 TEPPSGPSGL PLLPVLFALG GLLLSNASC VGGVLWQRRL RRLAEGISEK  
 1101 TEAGSEEDRV RNEYEESQWT GERDTQSSTV STTEAEPYR SLRDFSPQLP  
 1151 PTQEEVSYSR GFTGEDEDM FPGHLYDEVE RTYPPSGANG PLYDEVQMGF  
 1201 WDLHWPEDTY QDPRGIYDQV AGDLDTLEPD SLPFELRGHL V

FIG. 4A

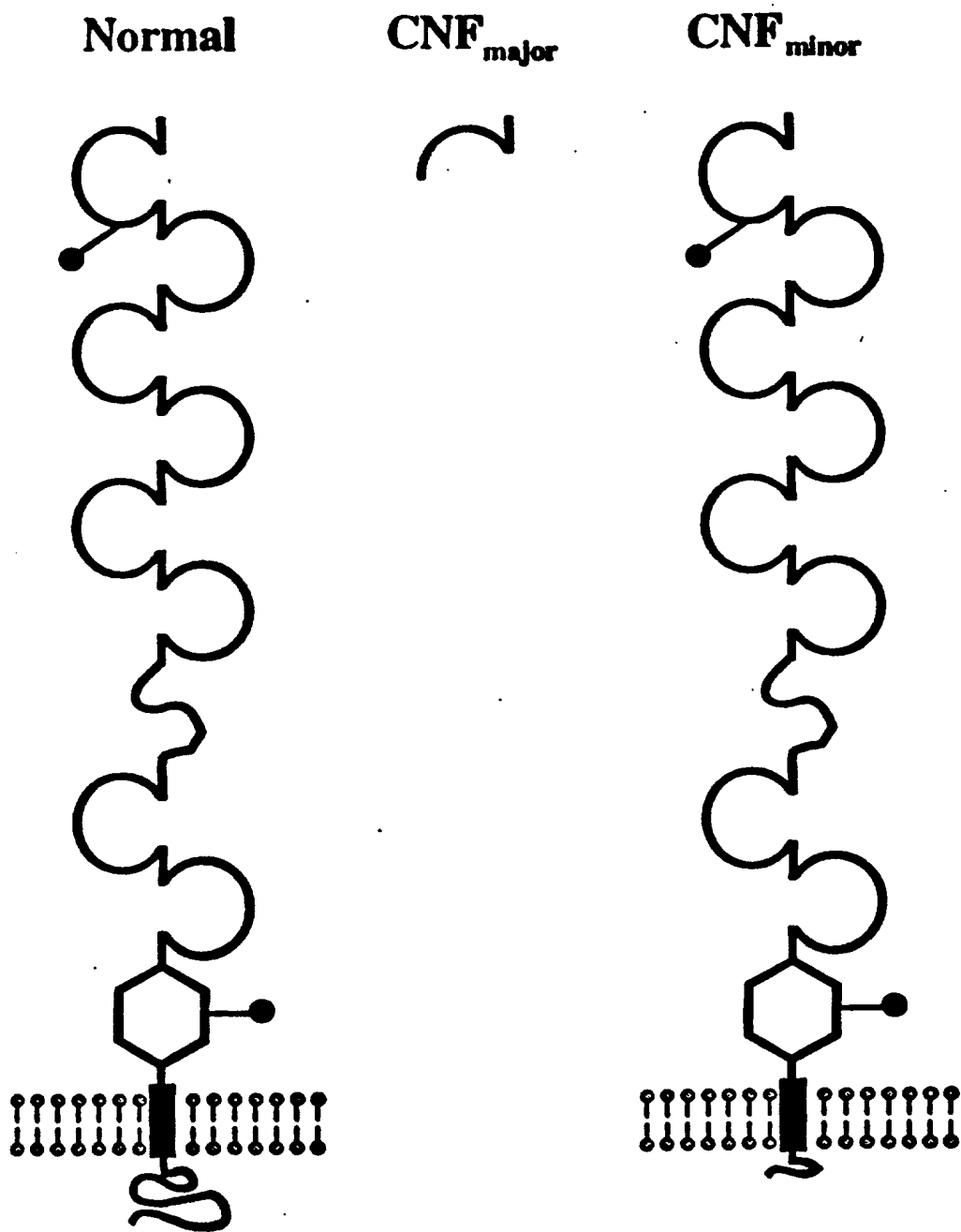


FIG. 4B

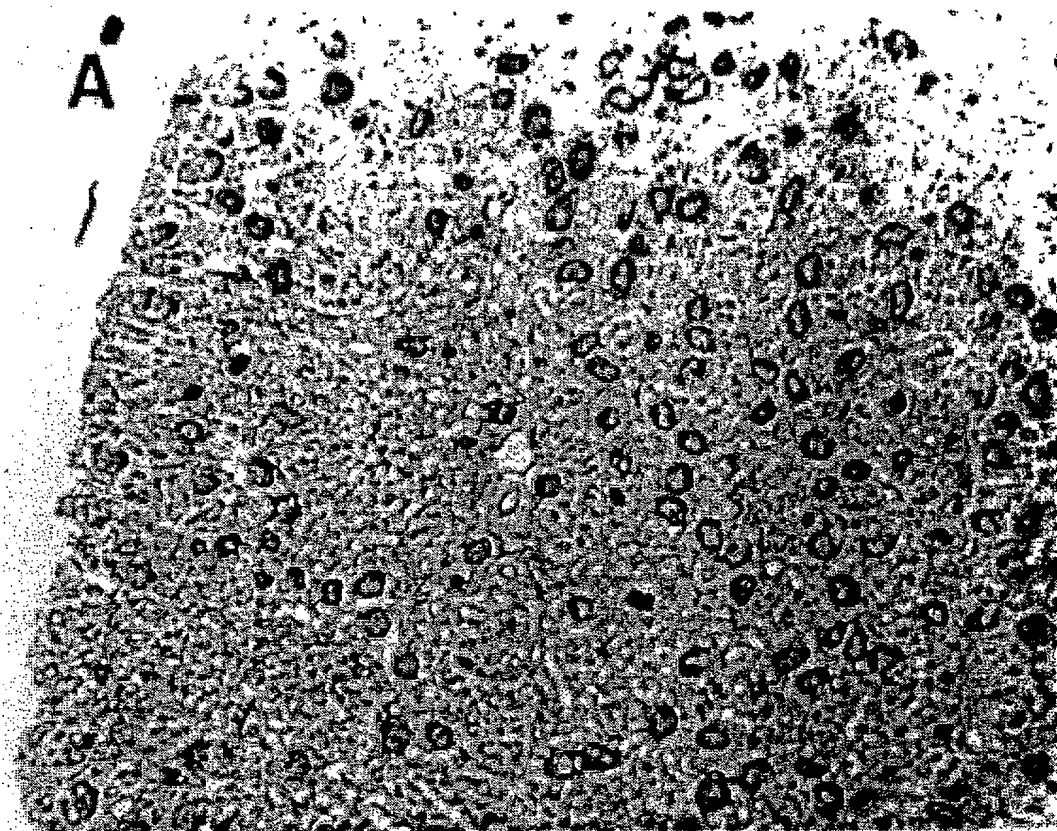
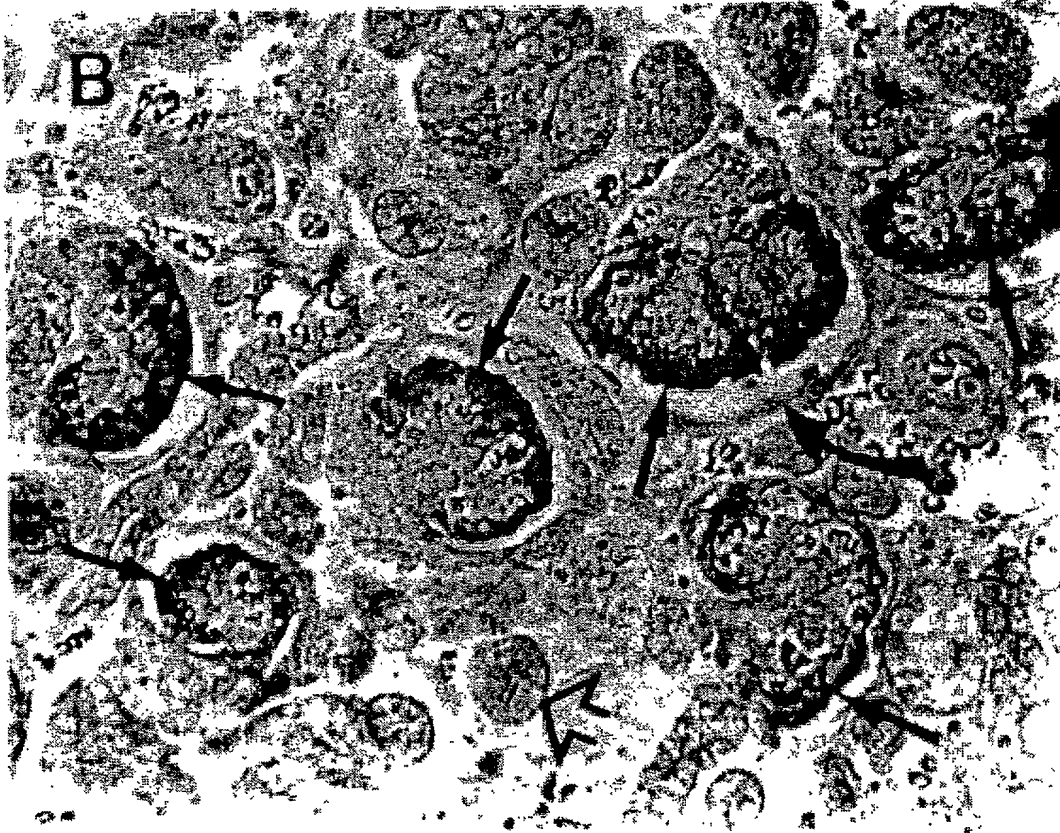


FIG. 5A



**FIG. 5B**